ALPL gene

alkaline phosphatase, liver/bone/kidney

Normal Function

The *ALPL* gene provides instructions for making an enzyme called alkaline phosphatase. This enzyme plays an important role in the growth and development of bones and teeth. It is also active in many other tissues, particularly in the liver and kidneys. This enzyme acts as a phosphatase, which means that it removes clusters of oxygen and phosphorus atoms (phosphate groups) from other molecules.

Alkaline phosphatase is essential for the process of mineralization, in which minerals such as calcium and phosphorus are deposited in developing bones and teeth. Mineralization is critical for the formation of bones that are strong and rigid and teeth that can withstand chewing and grinding.

Health Conditions Related to Genetic Changes

hypophosphatasia

More than 190 mutations in the *ALPL* gene have been identified in people with hypophosphatasia. About 80 percent of these mutations change a single protein building block (amino acid) in the alkaline phosphatase enzyme. Other mutations insert or delete genetic material in the *ALPL* gene or change the way the gene's instructions are used to build the enzyme.

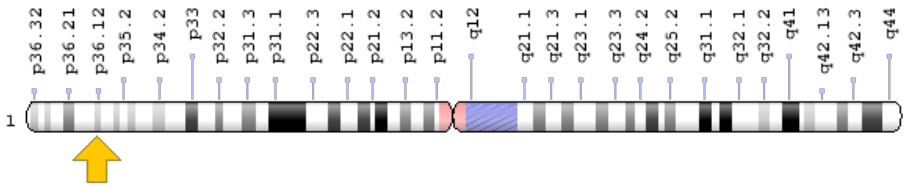
Mutations in the *ALPL* gene lead to the production of an abnormal version of alkaline phosphatase that cannot participate effectively in the mineralization of developing bones and teeth. A shortage of alkaline phosphatase allows substances that are normally processed by the enzyme to build up abnormally in the body. Researchers believe that a buildup of one of these compounds, inorganic pyrophosphate, underlies the defective mineralization of bones and teeth in people with hypophosphatasia.

ALPL mutations that almost completely eliminate the activity of alkaline phosphatase usually result in the more severe forms of hypophosphatasia. Other mutations, which reduce but do not eliminate the activity of the enzyme, are often responsible for milder forms of the condition.

Chromosomal Location

Cytogenetic Location: 1p36.12, which is the short (p) arm of chromosome 1 at position 36.12

Molecular Location: base pairs 21,508,982 to 21,578,412 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- alkaline phosphomonoesterase
- AP-TNAP
- glycerophosphatase
- HOPS
- MGC161443
- PPBT_HUMAN
- tissue non-specific alkaline phosphatase
- tissue-nonspecific ALP
- TNAP
- TNSALP

Additional Information & Resources

GeneReviews

- Hypophosphatasia
  https://www.ncbi.nlm.nih.gov/books/NBK1150

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ALPL%5BTIAB%5D%29+OR+%28alkaline+phosphatase+AND+hypophosphatasia%5BTIAB%5D%29%29+OR+%28%28tissue+nonspecific+alkaline+phosphatase%5BTIAB%5D%29+OR+%28tissue+non-specific+alkaline+phosphatase%5BTIAB%5D%29+OR+%28TNAP%5BTIAB%5D%29+OR+%28TNSALP%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- ALKALINE PHOSPHATASE, LIVER
  http://omim.org/entry/171760

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_ALPL.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=ALPL%5Bgene%5D
- HGNC Gene Family: Alkaline phosphatases
  http://www.genenames.org/cgi-bin/genefamilies/set/1072
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=438
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/249
- Tissue nonspecific alkaline phosphatase (ALPL) gene mutation database
  http://www.sesep.uvsq.fr/03_hypo_mutations.php
- UniProt
  http://www.uniprot.org/uniprot/P05186

Sources for This Summary

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